

What is claimed is:

1. A method of determining whether an individual has or is at risk for developing Usher syndrome Type IIa, the method comprising:
  - 5 obtaining a biological sample from the individual;  
incubating the biological sample with at least one antibody which is immunoreactive with at least a portion of a human usherin protein under conditions effective to produce an immunoconjugate if the usherin protein is present, wherein a complement of a polynucleotide encoding the usherin protein is capable of
  - 10 hybridizing to the polynucleotide represented by SEQ ID NO:3 under highly stringent hybridization conditions;  
evaluating for the presence or absence of the immunoconjugate; and  
correlating the presence of the immunoconjugate with the individual not having Usher syndrome Type IIa, and the absence of the immunoconjugate with
  - 15 the individual having or being at risk for developing Usher syndrome Type IIa.
2. The method of claim 1 wherein the biological sample is selected from the group consisting of at least a portion of testis, cochlea, epididymus, ovary, eye, uterus, heart, pancreas, prostate, skin, placenta, spleen, submaxillary gland, small
- 20 intestine, large intestine, blood vessels, and combinations thereof.
3. The method of claim 1 wherein the at least one antibody is detectably labeled.
- 25 4. The method of claim 3 wherein the detectable label is selected from the group consisting of radioactive labels, non-radioactive labels, and combinations thereof.

5. The method of claim 1 wherein the antibody is a monoclonal antibody, a polyclonal antibody, or combinations thereof.

5 6. The method of claim 1 wherein the antibody is immunoreactive with a polypeptide selected from the group consisting of SEQ ID NO:1, SEQ ID NO:2, SEQ ID NO:4, and combinations thereof.

7. The method of claim 1 wherein the polynucleotide encoding the usherin  
10 protein is represented by SEQ ID NO:3.

8. A method for detecting the presence or absence of an usherin protein, the method comprising:

obtaining a biological sample;  
15 incubating the biological sample with at least one antibody which is immunoreactive with at least a portion of a human usherin protein under conditions effective to produce an immunoconjugate if the usherin protein is present, wherein a complement of a polynucleotide encoding the usherin protein is capable of hybridizing to the polynucleotide represented by SEQ ID NO:3 under highly  
20 stringent hybridization conditions;  
evaluating for the presence or absence of the immunoconjugate;  
correlating the presence of the immunoconjugate with the presence of usherin protein, and the absence of the immunoconjugate with the absence of the usherin protein.

25 9. The method of claim 8 wherein the biological sample is selected from the group consisting of at least a portion of testis, cochlea, epididymus, ovary, eye, uterus, heart, pancreas, prostate, skin, placenta, spleen, submaxillary gland, small intestine, large intestine, blood vessels, and combinations thereof.

30

10. The method of claim 8 wherein the antibody is detectably labeled.
11. The method of claim 10 wherein the detectable label is selected from the  
5 group consisting of radioactive labels, non-radioactive labels, and combinations thereof.
12. The method of claim 8 wherein the antibody is a monoclonal antibody, polyclonal antibody, or combinations thereof.
- 10 13. The method of claim 8 wherein the antibody is immunoreactive with a polypeptide selected from the group consisting of SEQ ID NO:1, SEQ ID NO:2, SEQ ID NO:4, and combinations thereof.
- 15 14. The method of claim 8 wherein the polynucleotide encoding the usherin protein is represented by SEQ ID NO:3.
15. A method of determining whether an individual has or is at risk for developing Usher syndrome Type IIa, the method comprising:  
20 obtaining a biological sample from the individual;  
incubating the biological sample with a first antibody and a second antibody that are immunoreactive with at least a portion of a human usherin protein under conditions effective to produce an immunoconjugate if the usherin protein is present, wherein a complement of a polynucleotide encoding the usherin protein is  
25 capable of hybridizing to the polynucleotide represented by SEQ ID NO:3 under highly stringent hybridization conditions;  
evaluating for the presence or absence of the immunoconjugate; and  
correlating the presence of the immunoconjugate with the individual not having Usher syndrome Type IIa, and the absence of the immunoconjugate with  
30 the individual having or being at risk for developing Usher syndrome Type IIa.

16. The method of claim 15 wherein the immunoconjugate is a sandwich comprising the first antibody, the second antibody, and the human usherin protein.

5 17. The method of claim 15 wherein either the first antibody or the second antibody has an attached detectable label.

18. The method of claim 17 wherein the detectable label is selected from the group consisting of radioactive labels, non-radioactive labels, and combinations  
10 thereof.

19. The method of claim 15 wherein at least one of the first or second antibody is a monoclonal antibody.

15 20. The method of claim 15 wherein the first antibody is a monoclonal antibody attached to a solid surface and the second antibody is a polyclonal antibody with an attached detectable label.

21. The method of claim 20 wherein the detectable label is selected from the group consisting of radioactive labels, non-radioactive labels, and combinations  
20 thereof.

22. The method of claim 15 wherein the first or second antibody is immunoreactive with a polypeptide selected from the group consisting of SEQ ID  
25 NO:1, SEQ ID NO:2, SEQ ID NO:4, and combinations thereof.

23. The method of claim 15 wherein the polynucleotide encoding the usherin protein is represented by SEQ ID NO:3.

24. A test kit for detecting the presence or absence of Usher syndrome Type IIa in an individual comprising:

- an antibody that immunoreacts with at least a portion of a human usherin protein, wherein a complement of a polynucleotide encoding the usherin protein is capable of hybridizing to the polynucleotide represented by SEQ ID NO:3 under highly stringent hybridization conditions; and
- a detectably-labeled usherin protein.

25. The test kit of claim 24 wherein the antibody is a monoclonal antibody, a polyclonal antibody, or combinations thereof.

26. The test kit of claim 24 wherein the antibody is attached to a solid surface.

27. The test kit of claim 24 wherein the detectable label is selected from the group consisting of radioactive labels, non-radioactive labels, and combinations thereof.

28. The test kit of claim 24 wherein the antibody is immunoreactive with a polypeptide selected from the group consisting of SEQ ID NO:1, SEQ ID NO:2, SEQ ID NO:4, and combinations thereof.

29. The method of claim 24 wherein the polynucleotide encoding the usherin protein is represented by SEQ ID NO:3.

30. A test kit for detecting the presence or absence of Usher syndrome Type IIa in an individual comprising:

- a first antibody that immunoreacts with a portion of a human usherin protein; and

a second antibody that immunoreacts with a portion of a human usherin protein;

wherein a complement of a polynucleotide encoding the usherin protein is capable of hybridizing to the polynucleotide represented by SEQ ID NO:3  
5 under highly stringent hybridization conditions.

31. The test kit of claim 30 wherein either the first antibody or the second antibody has an attached detectable label.

10 32. The test kit of claim 31 wherein the detectable label is selected from the group consisting of radioactive labels, non-radioactive labels, and combinations thereof.

33. The test kit of claim 31 wherein at least one of the first or second antibody is  
15 a monoclonal antibody.

34. The test kit of claim 31 wherein the first antibody is a monoclonal antibody attached to a solid surface and the second antibody is a polyclonal antibody with an attached detectable label.

20 35. The test kit of claim 34 wherein the detectable label is selected from the group consisting of radioactive labels, non-radioactive labels, and combinations thereof.

25 36. The test kit of claim 31 wherein the first or second antibody is immunoreactive with a polypeptide selected from the group consisting of SEQ ID NO:1, SEQ ID NO:2, SEQ ID NO:4, and combinations thereof.

30 37. The test kit of claim 31 wherein the polynucleotide encoding the usherin protein is represented by SEQ ID NO:3.

38. An antibody that immunoreacts with at least a portion of human usherin protein under conditions effective to produce an immunoconjugate if the usherin protein is present, wherein the absence of an immunoconjugate correlates to the diagnosis of or the individual being at risk for developing Usher Type IIa syndrome, and wherein a complement of a polynucleotide encoding the usherin protein is capable of hybridizing to the polynucleotide represented by SEQ ID NO:3 under highly stringent hybridization conditions.

39. The antibody of claim 38 wherein the antibody is a monoclonal antibody, a polyclonal antibody, or combinations thereof.

40. The antibody according to claim 38 wherein the antibody is immunoreactive with a polypeptide selected from the group consisting of SEQ ID NO:1, SEQ ID NO:2, SEQ ID NO:4, and combinations thereof.

41. The antibody according to claim 38 wherein the polynucleotide encoding the usherin protein is represented by SEQ ID NO:3.